congenital sucrase-isomaltase deficiency

Congenital sucrase-isomaltase deficiency is a disorder that affects a person's ability to digest certain sugars. People with this condition cannot break down the sugars sucrose and maltose. Sucrose (a sugar found in fruits, and also known as table sugar) and maltose (the sugar found in grains) are called disaccharides because they are made of two simple sugars. Disaccharides are broken down into simple sugars during digestion. Sucrose is broken down into glucose and another simple sugar called fructose, and maltose is broken down into two glucose molecules. People with congenital sucrase-isomaltase deficiency cannot break down the sugars sucrose and maltose, and other compounds made from these sugar molecules (carbohydrates).

Congenital sucrase-isomaltase deficiency usually becomes apparent after an infant is weaned and starts to consume fruits, juices, and grains. After ingestion of sucrose or maltose, an affected child will typically experience stomach cramps, bloating, excess gas production, and diarrhea. These digestive problems can lead to failure to gain weight and grow at the expected rate (failure to thrive) and malnutrition. Most affected children are better able to tolerate sucrose and maltose as they get older.

Frequency

The prevalence of congenital sucrase-isomaltase deficiency is estimated to be 1 in 5,000 people of European descent. This condition is much more prevalent in the native populations of Greenland, Alaska, and Canada, where as many as 1 in 20 people may be affected.

Genetic Changes

Mutations in the *SI* gene cause congenital sucrase-isomaltase deficiency. The *SI* gene provides instructions for producing the enzyme sucrase-isomaltase. This enzyme is found in the small intestine and is responsible for breaking down sucrose and maltose into their simple sugar components. These simple sugars are then absorbed by the small intestine. Mutations that cause this condition alter the structure, disrupt the production, or impair the function of sucrase-isomaltase. These changes prevent the enzyme from breaking down sucrose and maltose, causing the intestinal discomfort seen in individuals with congenital sucrase-isomaltase deficiency.

Inheritance Pattern

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal

recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

Other Names for This Condition

- congenital sucrose intolerance
- congenital sucrose-isomaltose malabsorption
- CSID
- disaccharide intolerance I
- SI deficiency
- sucrase-isomaltase deficiency

Diagnosis & Management

Genetic Testing

- Genetic Testing Registry: Sucrase-isomaltase deficiency
  https://www.ncbi.nlm.nih.gov/gtr/conditions/C1283620/

Other Diagnosis and Management Resources

- MedlinePlus Encyclopedia: Abdominal bloating
  https://medlineplus.gov/ency/article/003123.htm
- MedlinePlus Encyclopedia: Inborn errors of metabolism
  https://medlineplus.gov/ency/article/002438.htm
- MedlinePlus Encyclopedia: Malabsorption
  https://medlineplus.gov/ency/article/000299.htm

General Information from MedlinePlus

- Diagnostic Tests
  https://medlineplus.gov/diagnostictests.html
- Drug Therapy
  https://medlineplus.gov/drugtherapy.html
- Genetic Counseling
  https://medlineplus.gov/geneticcounseling.html
- Palliative Care
  https://medlineplus.gov/palliativecare.html
- Surgery and Rehabilitation
  https://medlineplus.gov/surgeryandrehabilitation.html

Additional Information & Resources

MedlinePlus

- Encyclopedia: Abdominal bloating
  https://medlineplus.gov/ency/article/003123.htm
- Encyclopedia: Inborn errors of metabolism
  https://medlineplus.gov/ency/article/002438.htm
- Encyclopedia: Malabsorption
  https://medlineplus.gov/ency/article/000299.htm
- Health Topic: Diarrhea
  https://medlineplus.gov/diarrhea.html
- Health Topic: Malabsorption Syndromes
  https://medlineplus.gov/malabsorptionsyndromes.html

Genetic and Rare Diseases Information Center

- Congenital sucrase-isomaltase deficiency
  https://rarediseases.info.nih.gov/diseases/7710/congenital-sucrase-isomaltase-deficiency

Additional NIH Resources

- National Digestive Diseases Information Clearinghouse: Diarrhea
  https://www.niddk.nih.gov/health-information/digestive-diseases/diarrhea

Educational Resources

- Disease InfoSearch: Sucrase-isomaltase deficiency, congenital
  http://www.diseaseinfosearch.org/Sucrase-isomaltase+deficiency%2C+congenital/6919
- MalaCards: sucrase-isomaltase deficiency, congenital
  http://www.malacards.org/card/sucrase_isomaltase_deficiency_congenital
- Merck Manual for Healthcare Professionals: Carbohydrate Intolerance
  http://www.merckmanuals.com/professional/gastrointestinal-disorders/malabsorption-syndromes/carbohydrate-intolerance
- Orphanet: Congenital sucrase-isomaltase deficiency
  http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=35122
- The American College of Gastroenterology: Belching, Bloating, and Flatulence
  http://patients.gi.org/topics/belching-bloating-and-flatulence/

Patient Support and Advocacy Resources

- CLIMB: Children Living with Inherited Metabolic Diseases
  http://www.climb.org.uk/
- National Organization for Rare Disorders (NORD)
  https://rarediseases.org/rare-diseases/disaccharide-intolerance-i/

ClinicalTrials.gov

- ClinicalTrials.gov
  https://clinicaltrials.gov/ct2/results?cond=%22congenital+sucrase-isomaltase+deficiency%22

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28congenital+sucrase-isomaltase+deficiency%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D

OMIM

- SUCRASE-ISOMALTASE DEFICIENCY, CONGENITAL
  http://omim.org/entry/222900

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Reprinted from Genetics Home Reference:
https://ghr.nlm.nih.gov/condition/congenital-sucrase-isomaltase-deficiency

Reviewed: July 2008
Published: March 21, 2017

Lister Hill National Center for Biomedical Communications
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National Institutes of Health
Department of Health & Human Services